DHH gene

desert hedgehog

Normal Function

The *DHH* gene provides instructions for making a member of the hedgehog protein family. Hedgehog proteins are important for early development in many parts of the body. The protein produced from the *DHH* gene is believed to be involved in male sexual development and in the formation of the perineurium, the protective membrane around each bundle of fibers within a nerve.

Health Conditions Related to Genetic Changes

Swyer syndrome

DHH gene mutations have been identified in a small number of people with Swyer syndrome, a condition affecting sexual development also known as 46,XY complete gonadal dysgenesis or 46,XY pure gonadal dysgenesis. Affected individuals have two mutated copies of the *DHH* gene in each cell.

People usually have 46 chromosomes in each cell. Two of the 46 chromosomes, known as X and Y, are called sex chromosomes because they help determine whether a person will develop male or female sex characteristics. Girls and women typically have two X chromosomes (46,XX karyotype), and boys and men ordinarily have one X chromosome and one Y chromosome (46,XY karyotype).

Mutations in the *DHH* gene in people with Swyer syndrome affect the process of sexual differentiation, preventing affected individuals with a 46,XY karyotype from developing male gonads (testes) and causing them to develop female reproductive structures (a uterus and fallopian tubes).

other disorders

DHH gene mutations have been identified in people with 46,XY disorder of sex development, also known as partial gonadal dysgenesis. These individuals have one mutated *DHH* gene in each cell. They may have external genitalia that do not look clearly male or clearly female (ambiguous genitalia) or other changes in the genitals and reproductive organs.

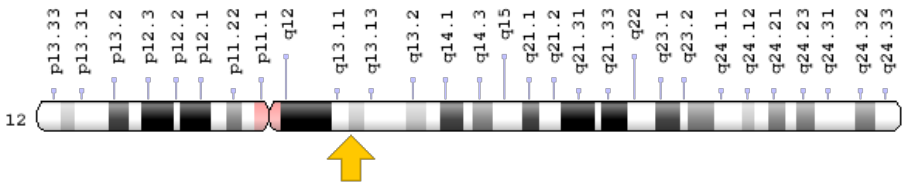
In addition to gonadal dysgenesis, some people with *DHH* mutations also have nerve abnormalities. These abnormalities affect nerves connecting the brain and spinal cord to muscles and sensory cells that detect sensations such as touch, pain, heat,

and sound (the peripheral nervous system). Affected individuals may experience weakness and loss of sensation in their extremities (peripheral neuropathy).

Chromosomal Location

Cytogenetic Location: 12q13.12, which is the long (q) arm of chromosome 12 at position 13.12

Molecular Location: base pairs 49,086,656 to 49,094,819 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- desert hedgehog homolog (Drosophila)
- DHH_HUMAN
- HHG-3
- MGC35145

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): The Hedgehog Family https://www.ncbi.nlm.nih.gov/books/NBK10071/#A1042
- Endotext: Sex Determination https://www.ncbi.nlm.nih.gov/books/NBK279001/#_sex-differentiation_toc-sex-determination_

GeneReviews

- Nonsyndromic Disorders of Testicular Development https://www.ncbi.nlm.nih.gov/books/NBK1547

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28DHH%5BTIAB%5D%29+OR+%28desert+hedgehog%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- 46,XY GONADAL DYSGENESIS, PARTIAL, WITH MINIFASCICULAR NEUROPATHY
  http://omim.org/entry/607080
- DESERT HEDGEHOG
  http://omim.org/entry/605423

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DHH.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DHH%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2865
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/50846
- UniProt
  http://www.uniprot.org/uniprot/O43323

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